CONGENITAL HYPOTHYROIDISM (CH)

(con-JEN-I-tal HIGH-poe-THIGH-royd-ism)

What is it?

Congenital hypothyroidism (CH) is a disease that is caused by the under activity of the thyroid gland. This under activity results in underproduction of thyroid hormones. These hormones play an important role in proper body metabolism. In growing children these hormones are essential for normal physical growth and mental development. Left untreated, this deficiency can result in mental retardation and stunted growth. Affected newborns may appear normal up to three months of age or have very subtle clinical symptoms. If detected early and maintained on treatment (hormone replacement medication) infants with CH should have normal growth and development.

How do you get it?

Congenital hypothyroidism occurs sporadically and is not usually an inherited disorder. The disorder is not associated with any prenatal lifestyle or risk factors. A more rare form of CH (about 15 percent of the cases) does involve an inborn (autosomal recessive) error in thyroid hormone synthesis.

Although the clinical signs of hypothyroidism may be subtle, infants with CH may exhibit some of the following symptoms: feeding problems, lethargy, prolonged postnatal jaundice, delayed stooling and constipation, enlarged protruding tongue, hoarse cry, protruding abdomen with an umbilical hernia, cold mottled skin, sluggish reflexes, patent posterior fontanelle with widely spread cranial sutures or delayed skeletal maturation for gestational age.

How common is it?

Congenital hypothyroidism occurs in 1 out of every 2,500 births in Missouri.

How is it treated?

Immediate diagnosis and treatment of congenital hypothyroidism in the neonatal period is critical to normal brain development and physical growth. Treatment is usually effective if started within the first few weeks of life. Delayed treatment may result in decreased intellectual capacity. Recommended treatment is lifetime daily administration of levo-thyroxine. Dosage will need to be gradually increased as the infant grows.

If your child needs additional testing or diagnostic evaluation, it is important that you follow through with the pediatrician's and/or specialist's recommendations for additional testing and referrals.

Treatment for primary congenital hypothyroidism will be life long.

Treatment is not curative and all morbidity cannot necessarily be prevented. Long-term management, monitoring and compliance with treatment recommendations are essential to the child's well being. A multidisciplinary approach is recommended and should include the following specialties: pediatrics and endocrinology.

Infants and children with congenital hypothyroidism should have regular follow-up appointments with a pediatric endocrinologist.

Periodic hearing evaluations also are recommended for children with CH, as hearing disorders are sometimes associated with congenital hypothyroidism.

Where can I get services?

Provision of the names below does not necessarily include all hospitals or private practice physicians who may treat children with CH.

Cardinal Glennon Memorial Hospital for Children
St. Louis, MO
314-577-5648

Children's Mercy Hospital
Kansas City, MO
816-234-3804

St. Louis Children's Hospital
St. Louis, MO
314-454-6051

University Hospital and Clinics
Columbia, MO
573-882-6979

What does DHSS offer?

State Public Health Laboratory – Newborn Screen

Related Links

Medline Plus (National Library of Medicine and the National Institutes of Health) www.medlineplus.gov

National Institutes of Health www.nih.gov

National Newborn Screening and Genetics Resource Center, http://genes-r-us.uthscsa.edu.

Organization for endocrine and metabolic disorders www.niddk.nih.gov

The MAGIC Foundation http://www.magicfoundation.org

GeneTests http://www.genetests.org